

Chapter 2

Autism Spectrum Disorder: Diagnostic Considerations

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Abstract DSM-5 is a departure from previous diagnostic formats. Changes in emphasis are to be found in the salience of selected key symptoms. A more significant departure are the modifiers that can be used to express severity of selected groups of symptoms. Additionally specifiers and associated disorders including medical and genetic disorders should be included as part of an overall diagnostic picture.

Keywords Diagnosis · Differential diagnosis · Symptoms

Autism as a stand-alone diagnostic entity is no more. Diagnostic and Statistical Manual 5 eliminates Autism as a discrete diagnostic entity and consistent with other neurodevelopmental disorders that exist within a severity range, DSM-5 creates a continuum. Autism spectrum Disorder (ASD) (American Psychiatric Association 2013; Volkmar and Reichow 2013). The essential ingredients of the original concept, now sixty years old, of Kanner's Autistic Disturbances of Affective Contact (Kanner 1943), deficits of social-emotional reciprocity, communication/play and the narrow interests of the individual with Autism remain as part of DSM-5. What distinguishes DSM-5 from its predecessor is the constriction of the richness of the menu of criteria (Dickerson Mayes et al. 2013). Importantly, the number of criteria was reduced from three to two, focusing upon social communication deficits and restricted, repetitive behaviors. Another change loosened the age of onset to the early developmental period from a disorder that must start before age three. Finally to better capture the focus of the range of severity the DSM-5 adds a symptom severity scale.

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2.1 DSM-IV Transitions to DSM-5

The category of pervasive developmental disorder (PDD-NOS) in DSM-IV included the previous separate disorders for which the following names were given: infantile autism, childhood autism, Kanner's autism, high-functioning autism, atypical autism, pervasive developmental disorder NOS, Childhood Disintegrative Disorder and Asperger's disorder (American Psychiatric Association 2000; Gibbs et al. 2012). The notion that these were discrete disorders began to unravel as the distinctions between PDD-NOS and for example, Asperger's eroded. Reflecting the porosity of boundaries and with the literature now replete with ASD studies, DSM-5 followed suit.

2.2 DSM-5 Criteria

DSM-5 has two main criteria sets: Diagnostic "A" criterion contains both deficits in social communication and social interaction. The disturbance in social relatedness includes marked impairment in non-verbal communication, peer relationships and social-emotional reciprocity. Impairments in communication include either a delay or total lack of spoken language (without an attempt to compensate through other means) or, for verbal individuals, a marked difficulty in the ability to sustain or initiate conversation, stereotyped and repetitive (or idiosyncratic) language and lack of developmentally-appropriate make-believe or social play. There are three items with descriptors around the items. The first of these: social-emotional reciprocity is conceived as a range from failure of the reciprocal behavior at one end of the range to a reduced quantity and quality of reciprocal behavior at the other end. The second item, nonverbal communicative behavior spans poor eye contact to the absence of nonverbal communication. Third is the area of social relationships. Here the range includes awkwardness of interaction to absence of peer interest. All three are required to fulfill "A" criterion. To capture the range of dysfunction for criterion "A" the severity scale from "1" denoting support required to and "3" substantial support such as the presence of an specialized caretaker should be applied.

Criterion B has four items that describe the behaviors of ASD. The first is repetitive motor stereotypies, repetitive use of objects, or repetitive speech. Echolalia, stereotypies and lining up toys are examples. The second focuses on routines and rituals. The behaviors could be verbal such as greeting rituals, or thinking patters such as expecting furniture to remain exactly in place or simply changing from one behavior to another as is required in transitioning from one play activity to another. The third is overvalued interest in an object, sensation or activity with a significant disinterest in other possible stimuli. Collecting every version of the same song for example shows the narrowness of the person's interest. Lastly is sensory abnormalities that can vary from hyper to hypo-reactive. The variety of behaviors associated with sensory experiences with, for example, water play that might be due to the

fascination with the temperature or the way light plays on it or the characteristics of liquids demonstrates the innumerable variations to be considered for this item. Compared to “A” criterion that requires every item to be satisfied, only two of these four items must be met for a total of six items. As with Criterion “A” a severity level should be applied that specifies whether low (level one) to high (level three) support is appropriate.

Criterion C changes the expected onset to be identified to the early developmental period as noted above. Criterion D like all disorders must be accompanied by clinically significant functional impairment. And finally, criterion E is the exclusion criterion that eliminate ASD if another disorder better describes the findings. The most common example of this would be global developmental delay which would include motor and/or sensory delays as part of the total presentation.

In addition to the specification of severity, other specifics are now part of the DSM-5 diagnostic package (McPartland et al. 2012). Instead of indicating cognitive impairment on Axis 2, the ASD diagnosis will include the presence or absence of accompanying intellectual impairment. DSM-5 has done away with the multiaxial system. The place for this common comorbid problem finds its way as a modifier of ASD. Although social communication can be severely delayed in ASD, a separate diagnostic item is no longer part of the criteria. Item 2 under criterion A is about nonverbal communication and echolalia which was part of verbal deviances in previous DSM editions and is no longer a main item and is found under repetitive movement’s item 1 criterion “B”. Hence it is important to specify whether ASD exists with or without a language impairment.

In the older multiaxial system a genetic or medical disorder associated with a diagnosis on axis one or two would be positioned under axis three. Where to place that information now? Another accessory to the diagnosis allows for a medical or genetic disorder or an environmental factor to be associated with ASD. ASD may be associated with Fragile X or with lead exposure or epilepsy.

When ASD is associated with other comorbidities they should be listed as associated disorders. Attention deficit hyperactivity disorder, Bipolar Disorder or Oppositional defiant disorder would be listed here.

2.3 Severity and Specificity in Practice

The consequences of adding the severity and specificity modifiers can create confusing complexity. A somewhat exaggerated example might look like: ASD accompanied by moderate intellectual disability with accompanying severe expressive language impairment and moderate receptive language impairment with very limited vocabulary with even greater limitations of communicative intent, with a genetic abnormality at 22q11.2 similar to velocardiofacial syndrome, with delivery at 28 weeks accompanied by anoxia without catatonia with level two severity of language and level 3 severity of restricted, repetitive behavior due to inability to tolerate routine transitions without extreme aggressive behavior. The extent of the

work required to obtain the information to quantify and describe all of these qualifiers is extensive and may be prohibitive in many community clinics as currently resourced.

Of special interest due to the proliferation of scientific literature on the subject, is the symptom cluster of catatonia. The specific criteria for catatonia, true for all the other associated comorbidities and conditions, must be met. If catatonic features are present, then ASD “with catatonia” as a modifier would be added.

2.4 Associated Features

Not unique to ASD but of interest diagnostically because it highlights special targets of treatment that further explains the features of the disorder in a particular patient are associated features (Volkmar et al. 2014). Associated features can be differentiated from comorbid disorders as they are aspects that are not uncommon but in and of themselves not disease states. Some associated features that have needed responses from the care delivery system include poor receptive language, and uneven IQ scores. These two deficits will need to be addressed especially in educational settings. Listing them alerts the school that special services will be necessary. A different intervention perhaps led by occupational or physical therapists may be necessary to work with motor deficits such as hypotonia or toe walking. Therefore, within the diagnostic language, these associated features could be mentioned. Finally, a behavior that influences quality of life, and perhaps impacts the restrictiveness of a community placement is self-injury. Listing this behavior as an associated feature should marshal the behavioral and medical resources that influence this problem.

2.5 Diagnostic Criteria Over Developmental Periods

While the criteria are static, the unfolding of symptoms of ASD over time is not. Development changes the picture of the prominence of the autistic features without straying from the core features of autism. For example adolescents with ASD may have very defined rituals and compulsive behaviors within their narrow range of interests. In contrast the preschool child with ASD may be notable for the failure of communicative interaction (Shattuck et al. 2007).

As ASD individuals reach legal age their care may shift to providers who are without training in the pediatric population. This has implications for diagnosis and the reliability of diagnosis in the adult age group. Some differences from the plethora of diagnostic symptoms seen in childhood will confront the adult practitioner. Older ASD individuals not only bear their own developmental symptom topologies but exhibit the impact of years of treatment interventions which may have led to compensations and skills so that their deficits may be masked by their life-long

experiences. A thorough review of history and childhood records will allow for a better understanding of the continuity of diagnosis over time.

Rett's syndrome: The reader may have noticed that Rett's syndrome, now a genetically identifiable disorder is no longer part of the ASD discussion. Rett's should be thought of perhaps like Fragile X with genetic abnormalities that affect brain function bringing it in phenotypic proximity to diagnostic criteria for ASD. For Rett's, a portion of the affected individuals life has symptoms that overlap with ASD. To capture this, the diagnosis might be stated as ASD with Rett's syndrome.

2.6 Differential Diagnoses

A number of psychiatric conditions have characteristics that overlap with symptoms of ASD. Rett's syndrome has been mentioned. Selective mutism however with significant expressive language handicap and over time social disabilities due to restricted social opportunities may bear some resemblance to ASD. Early social history and more typical behaviors in very specific settings can help to differentiate mutism from ASD.

A disorder with very pervasive effects on development, especially when manifested early in life is very early onset schizophrenia. Early onset schizophrenia is rare. When it occurs the available behaviors to analyze in the young child are few and developmental variation within a group of same age children can be wide. Obtaining evidence for abnormal perceptions such as auditory hallucinations requires extensive interactive interviewing skills and the ability to phrase questions that are relevant to the younger child. What will be clear is that the very young schizophrenic child needs help. The direction will be different if the condition is severe ASD. Gathering data from multiple sources and being open to the possibility of a diagnosis of very early onset schizophrenia rather than ASD improves the chance of proper diagnosis.

Another group of profoundly impaired children who may be mistaken for severe ASD are those with severe attachment disorder. Children exposed to a non-nurturing environment with unavailable attachment figures, experiencing sensory isolation of some duration and with little opportunity for normal social development will have characteristics of ASD. Motor stereotypies, language delay, and narrow range of interests, for example, willingness to eat only the few familiar foods that may have been available to them are prominent and similar to ASD features of early childhood. Differences however can include physical failure to thrive with significant growth curve retardation measures, rapid physical and psychological growth in resource-rich, nurturing settings and a history of neglect. Children from environments of significant deprivation present with poor language and social skills and improve as the environment facilitates these biologically hardwired potentials. The possibility that a child with ASD or other severe disability is at higher risk for neglect should be entertained.

Obsessive Compulsive Disorder (OCD) may be a differential diagnostic consideration when evaluating a child with odd behaviors. Overlap of specific compulsive behaviors such as touching stranger's body parts or certain objects, concern and agitation around changes to routines like bedtime rituals may meld with the variety of restricted and repetitive patterns of behavior such as lining up toys or flipping objects associated with ASD. Children who are well and then develop OCD symptoms that have its onset associated with Streptococcal infection should be tested for Pediatric Autoimmune Neuropsychiatric Disorder associated with Streptococcal infection (PANDAS). OCD behaviors with acute onset now called Pediatric acute-onset neuropsychiatric syndrome (PANS) should be considered as alternatives to a diagnosis of ASD.

Finally, a diagnostic category that impinges on the social deficits of ASD is Social Communication Disorder. The three main elements of this disorder are deficits in communication for social purposes, altering communication to fit the needs of the listener and some pragmatic aspects of language such as summarizing an experience, using verbal and nonverbal cues to improve communicative intent. While social communication is a major fixture for a diagnosis of ASD it is only one piece. ASD can be differentiated by examination of repetitive behaviors, narrow interests and other components of its diagnostic criteria.

2.7 Conclusion

The new emphasis for DSM-5 classification as related to diagnosis of ASD, has diminished the salience of sensory issues as well as the peculiarities of language of affected individuals. Echolalia as a symptom of social communication difficulties is now incorporated into repetitive behaviors. Language without a communicative message or issues of prosody are no longer criterion symptoms. The new DSM-5 orientation to a diagnosis of ASD is to add a method of scaling severity and connecting the disorder to the complex comorbidities and medical and genetic conditions that are so often associated with ASD. While it will take some effort to be able to fluidly express the extent of difficulties of an individual with ASD, the template has been posted.

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